Genomic Testing Results

A guide to understanding your test results and exploring your next steps

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★ **Sections that may have been personalized for you.**

How to use this booklet

- Some parts of this booklet have been filled in by your healthcare provider. This information can be found under the teal headers marked with a star (★), or in blue boxes.
- You will see some medical terms. These words are bolded, and are explained with in-text colour-coded definitions.
- If you are viewing the booklet electronically, you can click on the webpage links.

If you have any questions about your genetic testing results, please contact:

The Genomic Results Booklet was created by Julia Handra and the Friedman Lab, with the support of the CHILD-BRIGHT Network.

Results Summary ★

Date:
Name:

Click on the page number to jump to that section.

Genomic testing received: Whole Genome Sequencing (WGS)

Page 4

WGS reads all of your genetic instructions, also known as DNA. WGS looks for differences in the spelling of the DNA that may explain your health concerns. These differences are known as variants.

Who received testing? Trio: the person with the health condition and family members.

Genomic testing result: There has been a finding linked to your medical condition.

Impacted area or gene: Page 8

DNA Variant(s): Page 8

Related Medical Conditions: Page 8

Inheritance: Autosomal Recessive Page 9

Having two variants of this gene causes a health condition. Every child of this individual will inherit one variant, but it is unlikely they will have the same medical concerns.

Interpretation of the variant

Laboratory classification of the variant: Variant of Uncertain Significance (VUS) Page 6

The lab found a difference in DNA, but it is uncertain if this difference is responsible for a medical condition. There is currently not enough known to make a strong conclusion.

Doctor's Interpretation Page 7

Based on your medical history and your clinical picture, it is **very likely that** this is the cause of a medical condition.

Incidental (secondary) findings

Page 9

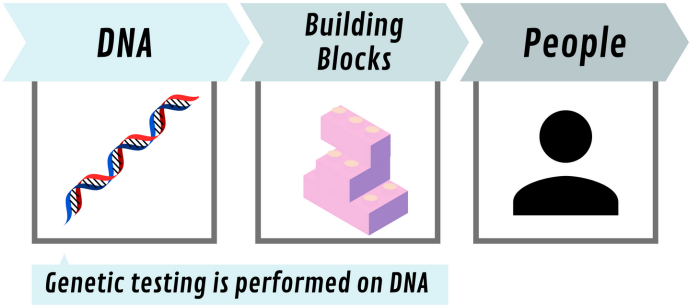
There were no incidental findings.

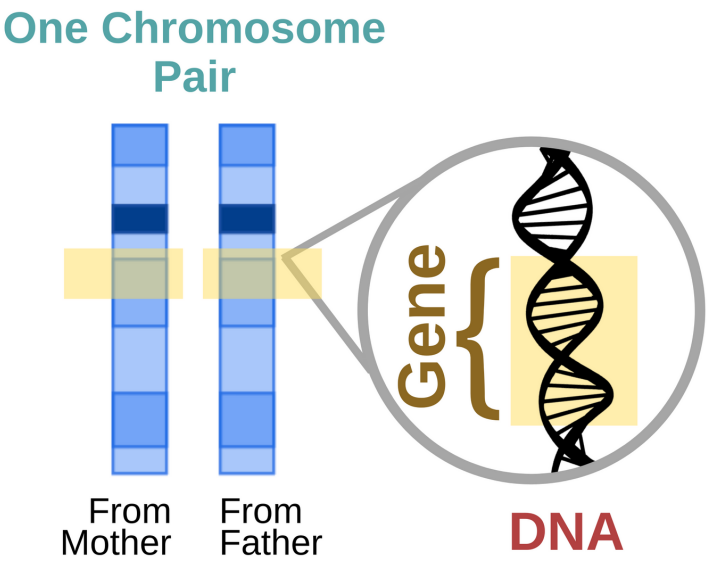
Notes from your healthcare provider

The rest of the booklet explains this information in more detail, and offers some resources.

Background Genomic Information

DNA is a long string of "letters."
Genes are the sections of your DNA that have instructions for making proteins.
Proteins are the building blocks of your body and play an important role in how you develop and function.





Genes are packaged in groups, called **chromosomes.** You have 23 **chromosomes** from your mother, and another matching 23 from your father.

Because **chromosomes** come in pairs, **genes** come in pairs too. You have two copies of each **gene**, one copy from each parent. This is why you have traits that are similar to those of your parents. The interaction between these two **gene** copies determines how a certain trait will show up.

Your unique set of DNA, including all of your genes, is called a **genome**. Your **genome** contains thousands of **genes**!

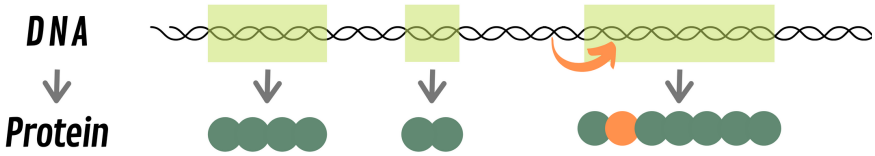
Over the past few decades, scientists have been working to identify and understand these **genes**. There is still a lot of work to be done. We don't fully know the role of every part of the **genome**, which is why there can be uncertainty in genetic testing results.

Your Notes

Genomic Testing

Differences in the DNA are known as **variants or mutations.** They occur frequently, and most are perfectly normal. Many account for traits that are shared between family members. However, sometimes a **variant** changes the instructions in a way that alters the **protein** product. This may cause a medical condition. Genetic technologies analyze **DNA** to find the **variants** that may cause medical concerns.

Testing You Received *



In the image above, the **DNA** that is highlighted in light green will be directly involved in making protein. This is called **protein-coding DNA**. There is still a lot of DNA in between. This is called **non-coding DNA**. **Non-coding DNA** can still have an impact on how proteins are made. For example, it may accelerate or slow down the creation of proteins.

Some tests look at all of the **DNA**, while others will only read the **protein-coding DNA**.

Your notes:

The genomic testing you received: Whole Genome Sequencing (WGS)

In relation to the image above, WGS will read all of the DNA, regardless of whether it is highlighted. This includes the DNA that does not directly code for proteins (non-coding DNA), but that can still have an impact on health. WGS can detect variants in both the protein-coding DNA and the non-coding DNA. This is why it is considered to be the most comprehensive and powerful genetic test available.

Who was tested?

Trio: the person with the health condition and family members.

Your Questions

Navigating genomic results is not something you have to do alone. Your doctors and/or genetic counsellors are there to support you through this process. It is our job to make sure that you understand what the genomic results mean for your family and support you through your next steps. We are best able to do so when we know more about your needs. You can help us understand these needs by asking us for the information that is most important to you.

Here are some questions patients often ask their doctor or genetic counsellor, and some space for you to keep track of your own questions.

Were you able to find a change in DNA that explains the symptoms?

Yes / Possibly

- What are these changes? Which genes are affected?
- How do these DNA changes impact health?
- Where did these changes in the DNA come from? Were they inherited?
- Is there anything we can change in our health management? Are there any other doctors we should see?
- Can this result change over time?

No

- Does these mean there isn't a genetic condition? Can this result change over time?
- If it isn't genetic, what else can explain these health concerns?
- What does this mean for our health management?

Your Genomic Testing Results ★

A snapshot of your testing process:

DNA Sample Collection → Genomic Testing & Analysis → Lab Results → Your Doctor's Interpretation

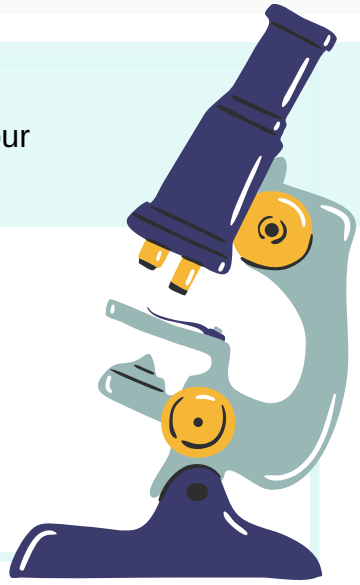
Genomic testing results can be complex and hard to understand. You might receive both laboratory results and a doctor's interpretation. These may seem different. Your doctor's interpretation is the most relevant to your care.

The lab looks for differences that may be causing health concerns. The lab classifies the DNA differences using computer tools and comparisons to other people's DNA. The lab does not have a full picture of patients' health concerns when making these classifications.

Your doctor interprets the lab results based on your health concerns. The doctor decides if these DNA differences are really affecting your health. Sometimes, no one can be certain what a difference in the DNA means. This uncertainty can be difficult.

Lab Results - Classification of the Variant(s) ★

Let's take a look at the lab results first. Remember, this is not based on your doctor's interpretation.



Variant of Uncertain Significance (VUS)

The lab found a difference in DNA, but it is uncertain if this difference is responsible for a medical condition. There is currently not enough known to make a strong conclusion.

Your notes:

Doctor's Interpretation: Is the DNA variant really causing the medical condition? ★

How does your doctor think the lab results fit into your medical picture?

During future consults, different specialists may interpret these results in their own way. Each specialist may have unique suggestions.

Based on your medical history and your clinical picture, it is **very likely that** this is the cause of a medical condition.

Are there any medical recommendations based on these results?

Fuzzy Endings

Genomic testing results are often a little "fuzzy" or uncertain. We are still learning to interpret all of the DNA variations that we see. We are working at the limits of our knowledge. **As our knowledge increases and genetic testing technology improves, the interpretation of the variant(s) might change.** You are encouraged to check back with your doctor from time to time to see if new information is available.

Here are some more details about the DNA variant(s) ★

Affected Gene(s):

What does this gene do in our bodies?

Variant(s):

Note that we cannot tell what caused these changes in the DNA.

Are there known medical conditions that have been associated with changes in this gene?

Your health concerns may not perfectly match the health conditions that have been previously reported.

Your notes:

Inheritance ★

Inheritance, or heredity, refers to how DNA and traits can be passed on from parent to child. We have two copies of each gene. One copy comes from the mother, and one copy comes from the father.

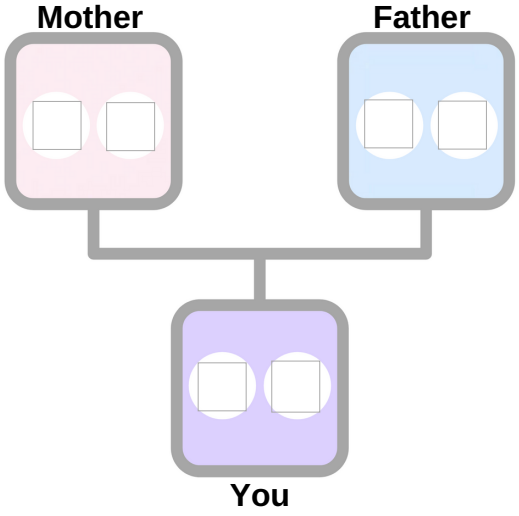
Because family members share some of the same DNA, they also share some of the same traits. Information about inheritance may be important in family planning. We can sometimes evaluate the chance of certain health conditions being passed on from parent to child.

This DNA variant is: Autosomal Recessive

An affected person has two variant copies of the gene. Having one variant and one normal copy is called being a 'carrier.' Being a carrier does not usually result in any effect on health. For an individual who has two variant copies, it is likely that one or both parents are carriers. All children of an affected individual will inherit a variant, but it is unlikely that they will have the same health concerns.

We have no control over our DNA, the variants that arise, and the variants we pass on.

Marks show which family members have the DNA variant(s).



Multiple family members were tested. Since the DNA of the mother, the father and the child was sequenced, we can identify all of the family members that have a certain variant.

Incidental (Secondary) Findings ★

There were no incidental findings.

While searching through your genome for variants that may be causing your health condition, we can seek and report additional health information. Such information is called an incidental (secondary) finding. Incidental findings are not related to the reason you received genetic testing, but can be important nonetheless. You have no incidental findings. This is not a guarantee of good health and you should still follow screening that your doctor recommends.

Next Steps

Sharing Your Results

Whether you share your results and who you share them with is **entirely your decision.** These are some people you may choose to share the results with.

Family:
Genetic information may be important for other family members. They may use this information for their own health or for family planning. Family and friends may be better able to help and support you if they can inform themselves about the genetic diagnosis. This can be a hard topic to discuss with others. Your doctor or genetic counsellor can offer guidance.

Your medical team – doctors, therapists, caretakers, counsellors, other healthcare providers:
A genetic diagnosis can change health management or help your healthcare team understand your medical condition. Only some of your doctors might have access to your genetic results. Therapists or caregivers likely do not. If you are unsure of how to explain your results to your healthcare team, you can share this booklet and the lab report with them.

Online platforms and genetic networks:
There are websites that connect families with similar genetic conditions. Posting information about your genetic results may also help to deepen the scientists' understanding and advance research. More information about sharing your results with the online community can be found under the Resources and Support section of this booklet. If you do choose to share this information online, it is important to check the privacy measures of the platform.

You can use this space to keep track of people you want to share your results with, or those with whom you have already shared results.

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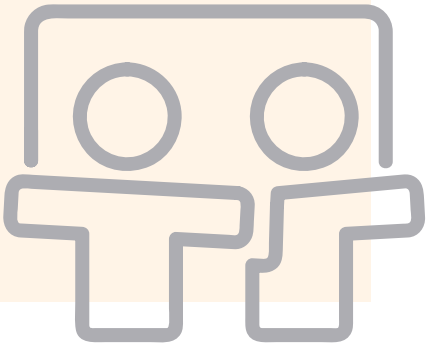


Resources and Support

Caring for your own health, or a family member's health, can be complex and challenging at times. While you may just be receiving a genetic diagnosis, we know that you have been putting in the hard work long before. You may already be well connected to resources that help you manage your medical concerns. Here are some other tips, websites and tools that may be helpful after receiving genetic testing results. These pages contain both general and case-specific resources.

Do not hesitate to reach out to your genetic counsellor or doctor if you need help in navigating options and resources.

When searching for resources, keep in mind that...

- Science literature can be very technical and hard to read. The way that papers are written may seem insensitive to patient-readers, or their family members. Patient websites are often more friendly when looking for information on a diagnosis.
- Each gene and every version of that gene can have very unique effects. This is why your medical concerns may be quite different from "similar" patients who are in medical publications. If you are doing online research, you might also see that different variants of the same gene can cause different conditions. Do not be alarmed. Focus only on the diagnosis that your doctor shared.
- Your family's needs may change over time.
- The best support caters towards symptoms and specific needs, as opposed to a diagnostic label.
- You are not alone. Many of the resources below connect families who are facing similar health concerns.

General Organizations and Alliances

☐ Global Genes

https://globalgenes.org/

There is a resource search tool. RARE List: A list of rare diseases, including an overview of the disease, support groups, news, events and clinical trials. RARE Portal: A platform that connects families.

“Empower patients, build communities & drive forward momentum for rare disease globally”
"The RARE Portal is your place to connect, find events and share your story."

☐ Disability Alliance BC (DABC)

http://disabilityalliancebc.org

Among other services, they can provide free legal advice to those with disabilities, and they offer assistance in applying for disability benefits.

"DABC’s Advocacy Access Program has been a place of support, information and one-to-one assistance for people with all disabilities."

To make an appointment, call Advocacy Access at **1-800-663-1278** (toll free).

☐ Family Support Institute of BC

https://familysupportbc.com

Support Worker Central: This database connects families with support workers in their area. FamilyWORKs is an initiative to create employment opportunities for people with disabilities.

"The purpose of the Family Support Institute of BC is to strengthen, connect and build communities and resources with families of people with disabilities in BC."

☐ Inclusion BC

https://inclusionbc.org

The Ready, Willing & Able initiative encourages employers to hire people with intellectual disabilities.

"Inclusion BC is a non-profit provincial organization that advocates for the rights and opportunities of people with intellectual disabilities and their families."

☐ PLAN: Planned Lifetime Advocacy Network

https://plan.ca/

PLAN, or Plan Institute in BC, focuses on creating support networks, tools for future planning, and community initiatives.

"PLAN is non-profit organization founded in 1989 to help families secure the future for loved ones with disabilities."

☐ Contact a Family

https://contact.org.uk

This website contains advice and support written by parents for parents, an online parent community forum, and patient-friendly medical information on genetics and various diagnoses.

“We support families with the best possible guidance and information. We bring families together to support each other. And we help families to campaign, volunteer and fundraise to improve life for themselves and others.

Resources for Rare Diseases

Rare Disease Foundation

https://rarediseasefoundation.org

This foundation has a Family Counselling Assistance Program and a Parent 2 Parent Resource Network. There are meetings and events in cities across Canada, including Vancouver.

Living Without a Diagnosis (a pamphlet):
https://rarediseasefoundation.org/wp-content/uploads/2017/07/Living-Without-a-Diagnosis.pdf

"The Rare Disease Foundation is focused on linking basic science and clinical practice to increase the efficiency of rare disease research. This model is called Translational Care. This model drives patient based, treatment focussed research projects from disease characterization to treatment with greater efficiency."

National Organization for Rare Disorders

https://rarediseases.org

There is an extensive database of rare diseases and their corresponding resources, along with advocacy and educational information. There are webinars on important topics for patients.

"Reports are written in patient-friendly language and each report links to disease-specific patient organizations and other resources that provide further support for patients and their families."

Rare Disease Information and Support Line

https://rqmo.org/rare-disease-information-and-resource-centre/

This is a support line you can call to receive help navigating a rare disorder. They can help you find more patient-friendly information and connect you with resources.

Toll-free number:
1-888-987-5539
Email: info@rqmo.org

This service is offered in English and French!

Organizations for Your Testing Outcome ★

Health Management Tools

Curatio *https://www.curatio.me/*
Curatio is an app that connects people with similar health conditions and helps to keep track of changes in health.

HealthVault, Medical Records, myPHR, Healthspek, My Medical *Free in your app store*
These smartphone apps allow you to track your medical history, store results from your medical tests, and share information with family or doctors. Make sure to check the security and privacy settings.

MyBooklet BC *https://mybookletbc.com*
This is a website that helps you design a personalized medical information booklet. This can help summarize medical histories and needs in a way that is easy to share with health care providers and support workers.

Scientific Information Sources

These sites have information for doctors and researchers, so the language can seem very complex. They show previous research and medical cases for a specific gene or diagnosis. Remember that different changes to a gene can produce different health conditions. When using these sites, make sure to look at the health condition that matches your testing results.

Website:	How to use it:
Genetics Home Reference - National Institute of Health (NIH)	This is a great source for information. The content is detailed but targeted to a patient audience. The site also offers educational content if you want to learn more about genetics. 1. Go to https://ghr.nlm.nih.gov/ 2. Using the text box in the upper right corner, type in the gene (from this booklet) or the health condition. 3. Once you are on the disease page, you can find sections on health management and other resources.
Your NIH link ★:	
Orphanet	Go to https://www.orpha.net/consor/cgi-bin/index.php Instructions can be found in this video: https://www.youtube.com/watch?v=57VPhtS4nME&t=
OMIM: Online Mendelian Inheritance in Man	Go to https://www.omim.org/ and enter the gene name in the search box. **OR** Look over your lab report (if you have received one) for an OMIM number (OMIM XXXXXX) and enter it into the search box.

Financial Resources and Subsidized Programs

Disability Tax Credit – *https://www.canada.ca/en/revenue-agency/services/tax/individuals/segments/tax-credits-deductions-persons-disabilities/disability-tax-credit.html*
"The disability tax credit (DTC) is a non-refundable tax credit that helps persons with disabilities or their supporting persons reduce the amount of income tax they may have to pay."

Child Disability Credit – *https://www.canada.ca/en/revenue-agency/services/child-family-benefits/child-disability-benefit.html*
"The child disability benefit is a tax-free monthly payment made to families who care for a child under age 18 with a severe and prolonged impairment in physical or mental functions."

Children and Youth with Special Needs (CYSN) Program BC
https://www2.gov.bc.ca/gov/content/health/managing-your-health/child-behaviour-development/special-needs

Registered Disability Savings Plan (RDSP) – *https://www.canada.ca/en/revenue-agency/services/tax/individuals/topics/registered-disability-savings-plan-rdsp.html*
"A registered disability savings plan (RDSP) is a savings plan that is intended to help parents and others save for the long term financial security of a person who is eligible for the disability tax credit (DTC)."
The government of Canada can contribute up to $3 for every $1 you put in.

Future Planning Tool by the Plan Institute – *https://futureplanningtool.ca/*
Build a plan to help you secure the future for you or anyone with a disability.

The Special Needs Planning Group – *http://www.specialneedsplanning.ca/index.html*
"This Web site is presented to you as a resource which will provide you with some basic information necessary to the understanding and implementation of plans for the future of your family member with a disability."
This parent-made website highlights legal and financial considerations for long term planning.

The At Home Program
https://www2.gov.bc.ca/gov/content/health/managing-your-health/child-behaviour-development/special-needs/complex-health-needs/at-home-program
"This program is intended to assist parents or guardians with some of the extraordinary costs of caring for a child with severe disabilities at home."

Nursing Support Services
http://www.bcchildrens.ca/our-services/sunny-hill-health-centre/our-services/nursing-support
"We are community-based registered nurses throughout BC who assist children and youth with medical complexities to live in their homes and in their communities."

Travel Assistance Program (for non-emergency medical services)
https://www2.gov.bc.ca/gov/content/health/accessing-health-care/tap-bc/travel-assistance-program-tap-bc

Finding a Support Group and Sharing Results With the International Community

People who have similar experiences can connect through support groups. These groups create a space for sharing experiences and feelings, building community, and learning from others. They can provide both emotional support and firsthand information. Support groups come in many different forms. Some are face-to-face while others are found on online platforms. They can be either private or open to the general public. Listed below are some platforms you may use in finding support groups. **Make sure to check the privacy and security conditions for each platform before sharing your information online.**

- **Facebook** fosters many local and international support groups. In the Facebook search bar, type [Disease Name] followed by [Location]. For example, "Cerebral Palsy Vancouver."
  - For rare diseases, try removing the location and adding "Disease". For example, "Cerebral Palsy Disease."
  - Some of these groups may be closed to the public in order to create a more private setting for the disease community. In this case, you will have to request to join by clicking "join group".
  - **You can also find more general support groups that are still helpful, supportive and informative. Try searching for "rare disease" or "complex kids" groups.**
- **Canadian Directory of National Support Groups**
  https://www.lhsc.on.ca/canadian-directory-of-genetic-support-groups/introduction-to-the-directory
  This directory will take you to a webpage specific to your health condition.
- **RareShare** https://rareshare.org/
  Create an account to join closed support groups specific to rare genetic conditions.
  - Here's a guide on how to get started: https://rareshare.org/howtos/welcome-to-rareshare
  - RareShare also has podcasts on specific conditions, as well as general podcasts on living with rare diseases.
- **RareConnect** https://www.rareconnect.org/en
  This user-friendly site fosters many online international communities for rare diseases. This site is available in 12 languages.
- **MyGene2** https://mygene2.org/MyGene2/
  Create an account to publicly share your variants with other families, researchers and clinicians. You can also search for families with variants of the same gene.
  - Here is a guide to the website: https://mygene2.org/MyGene2/downloadable/mygene2_flyer_families_2018-10-16.pdf

Recommended Support Groups ★

Genetic Databases

There are big genetic databases that allow scientists to share information and improve the understanding of genetic conditions. These databases are created using information from families who have received genetic testing.

Your doctor might ask you if would like to put your genetic results in a database. This usually means they will post details about your DNA and your health conditions. This will all be de-identified, so that there are no personal details. **Sharing your information is optional, and refusing to share will not impact your medical care.**

Pros of sharing:

- You can contribute to the pool of knowledge for your genetic condition.
- This information may help doctors find a diagnosis for patients with similar concerns.
- Many databases, like DECIPHER, are secure and not open to the public.

Cons of sharing:

- In some databases, researchers may not need to gain your consent before using genetic information that was posted.
- Some databases are easily accessed by the general public.

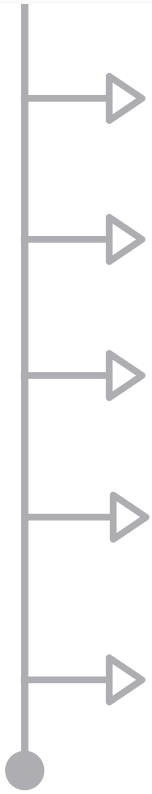
Be sure to check the terms of each database before sharing your genetic information!

Databases that contain your genetic information:

Extra notes from your healthcare provider ★

Your Next Steps

There is a lot of information to process and consider. Take your time. Here is some space to keep track of actions you would like to pursue. This may include connecting with a certain resource, visiting your family doctor, searching for a specialist, or any other changes in health management. **If you need support with this process, or still have questions about your results, please contact your genetic counsellor or doctor.**



This booklet is designed to be a supplement for genetic counselling, not a replacement for it. You can find a genetic counsellor in your area by using:

- **The Canadian Association of Genetic Counsellors Find a Clinic tool:** *https://www.cagc-accg.ca/?page=225*
- **The National Society of Genetic Counselors Find a Genetic Counselor tool:** *http://nsgc.org/p/cm/ld/fid=164*